PARENT FACT SHEET

DISORDER

Methylglutaric Aciduria (HMG)

CAUSE

HMG occurs when the HMG CoA lyase enzyme is missing or not working properly. This enzyme breaks down leucine, which is found in all foods that contain protein, and helps the body make ketones, used from fat stored in the body to produce energy. When a child is ill or goes without food too long, the body breaks down its own protein and fat to use for energy. Because of the deficient HMG CoA lyase enzyme, this can cause a metabolic crisis due to the HMG child's inability to process leucine and produce ketones.

IF NOT TREATED

HMG can exhibit different effects in each child. Babies with this condition are usually healthy at birth, although some show their first symptoms a few days after birth. Most babies, however, start to have symptoms between 3 months and 2 years of age. If not treated, many babies die during their first metabolic crisis. In surviving babies, repeated episodes of metabolic crisis can cause brain damage. This can result in life-long learning problems or mental delays.

TREATMENT OPTIONS

Your child will need to be under the care of a metabolic specialist and dietician. Treatment is usually needed throughout life.

- Your child needs to avoid going a long time without food. This is to avoid a metabolic crisis. These children should not go more than 4 to 6 hours without food and some may require more frequent feedings. It is important that these children be fed in the night – meaning you will need to wake them up to eat if they do not wake up on their own – and even if they are not hungry.
- A low-leucine diet with limited amounts of fat and protein and a high amount of carbohydrates is often recommended by the dietician in the form of special formula and foods.
- Some children may benefit from taking L-carnitine. This is a safe and natural substance that helps the body make energy. The metabolic specialist will decide if your child can benefit from this treatment. A prescription for this is required.
- Contact your child's doctor immediately at the start of any illness. Children with HMG need to be treated in a hospital to prevent serious health problems.

IF TREATED

With prompt and careful treatment, children with HMG lyase deficiency have a good chance to live healthy lives with typical growth and development. Even with treatment, some children still have repeated episodes of low-blood sugar and metabolic crisis. This can cause brain damage and may lead to life-long learning problems or mental delays.